ABSTRACT

Method for Detecting Growth Hormone Variations in Humans, the Variations and their Uses

A detection method for detecting a variation in *GH1* effective to act as an indicator of GH dysfunction in an individual, comprises the steps of comparing a test sample comprising a nucleotide sequence of the human *GH1* gene from the individual with a standard sequence known to be that of the human *GH1* gene. A difference between the test sample sequence and the standard sequence indicates the presence of a variation effective to act as an indicator of GH dysfunction (hereinafter "variant of *GH1*"). The test sample is obtained from a individual exhibiting the following criterion:

(i) growth failure, defined as a growth pattern [delineated by a series of height measurements; Brook CDG (Ed) Clinical Paediatric Endocrinology 3rd Ed, Chapter 9, p141 (1995, Blackwell Science)] which, when plotted on a standard height chart [Tanner *et al* Arch Dis Child 45 755-762 (1970)], predicts an adult height for the individual which is outside the individual's estimated target adult height range, the estimate being based upon the heights of the individual's parents.

Also disclosed are mutations thereby detected, and their use in screening patients for growth hormone irregularities or for producing variant proteins suitable for treating such irregularities.